

## Honors Biology Genetic Disease Research Project



### Introduction

Parents of a young family are seeking the help of a genetic research team to help save the lives of their children and possibly their future grandchildren. Genetic diseases claim the lives of millions of people each year and will affect generations down the line. The 21<sup>st</sup> century has brought us tremendous advances in medical treatment of these diseases. The so-called “DNA Revolution” has allowed the medical world to diagnose diseases in unborn babies, notify soon-to-be parents if they carry a diseased gene, and offer treatment options including fixing the defective gene. The parents need YOU to assist the genetic research team.

### The Task

Your task will be to present accurate information to parents about a specific disease which afflicts their children. As a member of this genetic research team, you will describe the genetic disorder and how it is diagnosed, explain how the condition can be treated (or cured) and present any ethical issues that may be involved in the treatment of this disease. Your research paper and presentation should thoroughly answer the following questions:

1. How does this genetic disorder occur? (Is it a sex-linked, autosomal, recessive or dominant trait?)
2. What type of mutation causes the disease?
3. Find out which chromosome the disease occurs on.
4. Determine the frequency of occurrence of the disorder in the whole population and specific ethnic groups.
5. Describe risk factors for inheriting the disorder.
6. What are the effects of the disease/symptoms of the disease?
7. Give the names of any famous people/historical figures that had/have the disease.
8. What techniques are used for prenatal diagnosis of the disease?
9. What are the treatments of the disease (if any)? Is the disease curable?
10. Are there any ethical issues involved with the treatment or diagnosis of the disease?

### The Paper

Each individual will turn in one 2 page, typed paper on the day of their presentation. **The paper should be double-spaced, 12-point font, Times New Roman, with 1 inch margins all around.** Points will be deducted if you do not follow these guidelines. All information should be in your OWN WORDS. I will not accept papers that have been copied from the internet or other sources. The paper should have the following format:

- I. Intro/Description (include historical figures/famous people here)
- II. Symptoms/Effects
- III. Diagnosis/Treatment
- IV. Causes
- V. Occurrence in population/ethnic groups
- VI. Ethical Issues/Conclusion

### **The Presentation**

Each individual must give a 3 – 5 minute presentation on their genetic disorder. Your presentation should include the same information given in the written paper, but do not just read the paper. Make your presentation interesting and original. You must include at least two visual aids. This can be in the form of posters, pictures, etc. OR you can do a PowerPoint presentation (this will count as 2 visual aids). Refer to the rubric below to see how the project will be graded.

### **The Rubric**

Research Paper:

- Follows specific criteria for format (12 point font, Times New Roman, double spaced, 1 inch margins) -**10 points**
- Length (2 full pages...no more than 5 pages)-**10 points**
- Intro/Description (include historical figures/famous people here)-**10 points**
- Symptoms/Effects-**10 points**
- Diagnosis/Treatment-**10 points**
- Causes-**10 points**
- Occurrence in population/ethnic groups-**10 points**
- Ethical Issues/Conclusion-**10 points**
- **Plagiarism will result in a loss of 75 points!!!**

You will receive full credit for each portion of the paper if it is thorough in explaining and describing the disease. You may label each section of your paper with the appropriate heading, but DO NOT create extra spaces between these headings and the paragraph below.

Presentation:

- Stays within time constraints of 3-5 minutes- **5 points**
- Presentation includes at least 2 visual aids OR PowerPoint that are appropriate to the presentation- **5 points**
- Presentation gives information on all aspects of the disease to be covered in the research paper-**5 points**
- Presenters speak loudly and clearly- **5 points**

Your presentation will receive full credit if it follows the guidelines listed above. I suggest that you “rehearse” your presentation at least once to make sure you can **PRONOUNCE** all of the terms used and to ensure you stay within the time constraints.